



Marfan syndrome is a genetic condition that affects connective tissue. Connective tissue is a type of tissue that helps to hold everything together, like a glue for your body. The main body systems affected by Marfan syndrome include the eyes (ocular), bone and joints (skeletal), and heart and blood vessels ([cardiovascular](#)).

- **Ocular:** The most common eye findings are severe nearsightedness (also called [myopia](#)) and displacement of the lens of the eye (also called ectopia lentis). Individuals with Marfan syndrome may also be at risk for early [cataracts](#), early [glaucoma](#), and [retinal detachment](#).
- **Skeletal:** Common findings include long arms and legs, long fingers and toes (also called arachnodactyly), a tall and thin body type, curvature of the spine (scoliosis), chest wall abnormalities (called pectus excavatum or carinatum), flat feet, high palate, and joint laxity (overly flexible).
- **Cardiovascular:** Differences with the heart and major blood vessels of the body are leading causes of serious health problems for people with Marfan syndrome. One of the biggest complications is called dilation of the aorta, which is the stretching of the main blood vessel that carries blood from the heart to the body. Dilation of the aorta can lead to an [aneurysm](#) (bulge in the aorta) or [dissection](#) (tear in the aorta). Both aortic [aneurysms](#) and [dissections](#) can be life threatening. Another heart problem, called [mitral valve prolapse](#) (MVP), is also more common in people with Marfan syndrome.

Additional things that can be seen in association with Marfan syndrome include collapsed lung (also called spontaneous pneumothorax), enlargement of the membrane around the brain and spinal cord (also called dural ectasia) which can cause pain, respiratory problems such as asthma or emphysema, stretch marks, and other health concerns. Some individuals may have learning difficulties, but this is rare.

Marfan syndrome can range in severity, and there can be variability within a family. Individuals may have few or no symptoms while others may have severe and early-onset health concerns.

### Causes

Marfan syndrome is caused by pathogenic variants in the *FBN1* gene, which are inherited in an [autosomal dominant](#) pattern. This means that a single copy of the pathogenic variant is enough cause an individual to develop the condition, and anyone who carries the pathogenic variant has a 50% chance to pass it down to any children they have. About 75% of people who have Marfan syndrome have a parent who also has it. The other 25% are the first ones



in their family to have it (called de novo). Marfan syndrome affects 1 in every 5,000-10,000 people.

### **Diagnosing Marfan syndrome**

While genetic testing for Marfan syndrome can be helpful, medical providers may use other pieces of information, such as a physical exam and family history, to help establish the diagnosis. Some red flags that can increase the chance for Marfan syndrome in a family include:

- Dilation of the aorta
- [Ectopia lentis](#) (dislocation of the lens of the eye)
- [Systemic score](#) (takes into account skeletal and other features)
- [Family history](#) of health concerns related to Marfan syndrome, or a family history of a pathogenic variant in the *FBN1* gene.

These criteria are called the “Ghent nosology” and the [Marfan Foundation](#) has created an online tool that incorporates these criteria.

Due to the varying types of genetic changes that have been reported in the *FBN1* gene in association with Marfan syndrome, genetic testing for Marfan syndrome involves [sequence](#) and [deletion/duplication](#) analysis. About 70-95% of those who meet the criteria for Marfan syndrome based on their personal and family history will have a pathogenic variant in the *FBN1* gene.

### **Medical Management for Marfan syndrome**

Treatment for Marfan syndrome can sometimes vary depending on the individual person and their specific health concerns, and should be discussed with a medical provider who is familiar with Marfan syndrome. Management for Marfan syndrome is generally focused on monitoring for the potential health complications. Individuals with Marfan syndrome are recommended to have regular eye exams by an ophthalmologist, annual cardiac imaging to monitor the aorta (screening may be more frequent if dilation is found), and intervention and follow-up by an orthopedist for those with progressive scoliosis or other skeletal problems. Additional screening may be recommended depending on the individual’s personal and [family history](#). Because of the [cardiovascular](#) and other risks, it is often suggested that individuals with Marfan syndrome avoid certain activities such as contact and competitive sports. Because of the potential risks, it is important for women with Marfan syndrome who are considering pregnancy to discuss options and review these risks



beforehand when possible.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

### **Resources**

[National Marfan Foundation](#)

[Genetic Aortic Disorders Association \(GADA\) of Canada](#)